

This listing of claims will replace all prior versions and listings of claims in the instant application:

Listing of Claims:

1-43 (cancelled)

44. (new) An oligonucleotide for detecting the presence of a mutation associated with Familial Dysautonomia, wherein said oligonucleotide comprises position 34,201 of SEQ ID NO:1 and wherein the thymine nucleotide at position 34,201 is replaced by a cytosine nucleotide.

45. (new) The oligonucleotide of claim 44 wherein the oligonucleotide comprises SEQ ID NO:85.

46. (new) An oligonucleotide for detecting the presence of a mutation associated with Familial Dysautonomia, wherein said oligonucleotide comprises position 33,714 of SEQ ID NO:1 and wherein the guanine nucleotide at position 33,714 is replaced by a cytosine nucleotide.

47. (new) The oligonucleotide of claim 46 wherein the oligonucleotide comprises SEQ ID NO:86.

48. (new) An oligonucleotide for detecting the presence of a mutation associated with Familial Dysautonomia, wherein said oligonucleotide comprises position 2,397 of SEQ ID NO:2 and wherein the guanine nucleotide at position 2,397 is replaced by a cytosine nucleotide.

49. (new) The oligonucleotide of claim 48 wherein the oligonucleotide comprises SEQ ID NO:86.

50. (new) An oligonucleotide consisting of 16 or more nucleotides of SEQ ID NO:1, wherein said oligonucleotide comprises position 34,201 of SEQ ID NO:1.

51. (new) An isolated and purified nucleic acid molecule for diagnosing Familial Dysautonomia in an individual by detecting a deletion of exon 20 in IKAP cDNA, wherein said exon 20 is nucleotides 2,441-2,514 of SEQ ID NO:2.

52. (new) The nucleic acid molecule of claim 51, wherein said nucleic acid molecule is an oligonucleotide which detects the nucleic acids sequence of exon 19 directly attached to the nucleic acid sequence of exon 21 in IKAP cDNA.

53. (new) The nucleic acid of claim 52, wherein said oligonucleotide comprises TACAGACTTA.

54. (new) An isolated and purified nucleic acid molecule consisting of a nucleic acid sequence selected from the group consisting of:

- (a) nucleotides 311-4,309 of SEQ ID NO:2, wherein the guanine nucleotide at position 2,397 is replaced by a cytosine nucleotide; and
- (b) SEQ ID NO:2, wherein the guanine nucleotide at position 2,397 is replaced by a cytosine nucleotide.

55. (new) A recombinant vector comprising a nucleic acid molecule according to any one of claims 44, 46, 48, 50, 51, or 54.

56. (new) The recombinant vector according to claim 55, wherein said nucleic acid molecule is operably linked to an expression control sequence suitable for expression of said nucleic acid sequence in a host cell.

57. (new) A host cell comprising the recombinant vector according to claim 55, wherein said host cell is selected from a group consisting of bacteria, fungi, insect cells, plant cells, or mammalian cells.

58. (new) The host cell of claim 57, wherein said bacteria is selected from the group consisting of *E. coli*, *Pseudomonas*, *Bacillus subtilis*, and *Bacillus stearothermophilus*.

59. (new) The host cell of claim 57, wherein said fungi is yeast.

60. (new) The host cell of claim 57, wherein said mammalian cells are selected from the group consisting of murine, bovine, porcine, and human.

61. (new) A kit for assaying for the presence of a mutation associated with Familial Dysautonomia in an individual comprising primers capable of amplifying a region of IKAP of sufficient size to detect a FD1 mutation or a FD2 mutation, wherein said region amplified comprises a FD1 or a FD2 mutation.

62. (new) The kit of claim 61, wherein said region amplified consists essentially of 500 nucleotides.

63. (new) The kit of claim 62, wherein said primers are 18F (SEQ ID NO:82) and 23R (SEQ ID NO:84).

64. (new) The kit of claim 61, wherein said region amplified consists essentially of 399 nucleotides.

65. (new) The kit of claim 64, wherein said primers are 19F (SEQ ID NO:83) and 23R (SEQ ID NO:84).

66. (new) The kit of claim 61, wherein the region amplified comprises position 33,714 of SEQ ID NO:1.

67. (new) The kit of claim 61, wherein the region amplified comprises position 34,201 of SEQ ID NO:1.

68. (new) The kit of claim 61, wherein the region amplified comprises position 2,397 of SEQ ID NO:2.

69. (new) A kit for assaying for the presence of a mutation associated with Familial Dysautonomia in an individual comprising at least one oligonucleotide probe which detects either a FD1 mutation or a FD2 mutation, wherein said probe comprises SEQ ID NO:85 or SEQ ID NO:86.

70. (new) A kit for assaying for the presence of a mutation associated with Familial Dysautonomia in an individual comprising at least one oligonucleotide probe which detects a deletion of exon 20 in IKAP cDNA, wherein said exon is nucleotides 2,441-2,514 of SEQ ID NO:2.

71. (new) The kit of claim 70 wherein said probe detects the nucleic acids sequence of exon 19 directly attached to the nucleic acid sequence of exon 21 in IKAP cDNA.

72. (new) The kit of claim 71 wherein said probe comprises TACAGACTTA.

73. (new) The kit according to any one of claims 61, 69, or 70 further comprising one or more oligonucleotide probes which hybridize to one or more additional genes, wherein said additional gene is associated with an additional genetic disorder and wherein said one or more oligonucleotide probes detects the additional genetic disorder.

74. (new) The kit according to claim 71, wherein the additional genetic disease is selected from the group consisting of Canavan's disease, Tay-Sachs disease, Goucher disease, Cystic Fibrosis, Fanconi anemia, and Bloom syndrome.

75. (new) A method of producing a mutant IKAP polypeptide, comprising:

(a) culturing a host cell transformed with a vector containing a DNA molecule encoding a mutant IKAP polypeptide in a cell culture medium under conditions whereby the mutant IKAP polypeptide is expressed, and

(b) isolating the thus-produced mutant IKAP polypeptide,
wherein said DNA molecule consisting of a nucleic acid sequence selected from the group consisting of SEQ ID NO:2 wherein the guanine nucleotide at position 2,397 is replaced by a cytosine nucleotide and nucleic acids 311-4,309 of SEQ ID NO:2 wherein the guanine nucleotide at position 2,397 is replaced by a cytosine nucleotide.